PHENYLKETONURIA (PKU)

(FEE-nil-Kee-tone-u-ree-ah)

What is it?

Phenylketonuria (PKU) is the result of an inability to break down the amino acid, phenylalanine, which is found in the protein of foods. It is a disorder of amino acid metabolism that results in excessive levels of phenylalanine in body fluids. Elevated levels of phenylalanine can become neurotoxic resulting in various degrees of mental retardation; early detection and treatment of hyperphenylalaninemia is necessary to prevent mental retardation.

How do you get it?

These disorders are inherited in an autosomal recessive pattern. As an autosomal recessive disorder, the parents of a child with PKU are unaffected. Healthy carriers of the condition have one normal and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene, resulting in PKU. Carrier parents have a 50 percent chance of having a child who is an unaffected carrier, such as both of the parents, and a 25 percent chance of having an unaffected, non-carrier child. These risks hold true for each pregnancy. All siblings of infants diagnosed with PKU or hyperphenylalaninemia.

Infants with PKU usually appear normal at birth. Early symptoms may include skin rash, seizures, excessive restlessness, irritable behavior and a musty odor of the body or urine. Later signs include developmental delays, gait disturbances and mental retardation.

How common is it?

PKU occurs in 1 out of every 15,000 births in Missouri.

How is it treated?

Early diagnosis and treatment through diet is essential to prevent developmental delays. Phenylalanine is an essential amino acid, and individuals with PKU require careful dietary management and monitoring for life. Individuals with PKU require a low phenylalanine diet, which consists of a specialized medical formula in combination with regular foods that a specialized medical formula (link to the Metabolic Formula Program) in combination with regular foods that are low in phenylalanine. There are

several low phenylalanine products available for use as formula for infants and as dietary supplements for older children and adults.

Women of childbearing age who are diagnosed with PKU or hyperphenylalaninemia require strict dietary control prior to conception and throughout pregnancy to reduce risk of complications, including miscarriage or of having an infant with severe birth defects due to high maternal levels of phenylalanine.

If your child needs additional testing or diagnostic evaluation, it is important that you follow through with the pediatrician's and/or specialist's recommendations for additional testing and referrals.

Treatment is life long and compliance with dietary management is imperative to your child’s health, growth and development.

Infants and children with PKU or hyperphenylalaninemia should have regular follow-up appointments with a metabolic disease specialist.

Treatment is not curative and all morbidity cannot necessarily be prevented.

Long-term management, monitoring, and compliance with treatment recommendations are essential to the child’s well being. A multidisciplinary approach is recommended and should include the following specialties: pediatrics, genetics and nutrition.

Where can I get services?

Cardinal Glennon Memorial Hospital for Children
St. Louis, MO
314-577-5639

Children’s Mercy Hospital
Kansas City, MO
816-234-3804

St. Louis Children’s Hospital
St. Louis, MO
314-454-6051

University Hospital and Clinics
Columbia, MO
573-882-6979

What does DHSS offer?

Metabolic Formula Program Fact Sheet

Related Links

Medline Plus (National Library of Medicine and the National Institute of Health) www.medlineplus.gov

National Coalition for PKU and Allied Disorders www.pku-allieddisorders.org

National Institutes of Health www.nih.gov

National Newborn Screening and Genetics Resource Center, http://genes-r-us.uthscsa.edu

GeneTests http://www.genetests.org